

MEN1 gene

Associated Syndrome Name: Multiple endocrine neoplasia type 1 (MEN Type 1)

MEN1 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Endocrine	High Risk
Other	High Risk

MEN1 gene Overview

Multiple endocrine neoplasia type 1 (MEN Type 1)^{1, 2, 3, 4}

- Individuals with *MEN1* gene mutations have multiple endocrine neoplasia type 1 (MEN Type 1).
- Patients with MEN Type 1 have a high risk for many different types of endocrine and non-endocrine tumors, which can occur at young ages. These tumors cause health problems when they become large, and/or when they secrete hormones. Some of these tumors can also become malignant (cancer).
- The most common type of tumor in people with MEN Type 1 is of the parathyroid gland. These occur in most people between age 20 and 25, and in almost everyone by age 50. These tumors lead to too much calcium in the blood (hypercalcemia), which can cause serious health issues affecting mental health (depression and confusion), the heart, the digestive system, the kidneys and bones.
- Other common cancers that occur in people with MEN type 1 include pituitary tumors and neuroendocrine tumors of the Gastro-Entero-Pancreatic (GEP) tract (e.g. pancreatic neuroendocrine tumors). Tumors can also develop in the small intestine, thymus, and lungs. Some of these have a high risk of becoming malignant (cancerous).
- Many people with MEN Type 1 will have non-cancerous tumors of the skin (angiofibromas and collagenomas), fatty tissue (lipomas), soft muscle (leiomyomas, also known as fibroids) and the central nervous system (ependyomas and meningiomas). These tumors require treatment if their size or location causes health problems.
- Some studies have described a possible increased risk for breast cancer in individuals with *MEN1* mutations. However, these studies are not conclusive and there are currently no medical management guidelines to address these possible risks.
- While individuals with MEN Type 1 have a high risk for cancers and other serious health concerns, these risks can be greatly reduced with appropriate medical management. A limited summary of detailed guidelines from the National Comprehensive Cancer Network (NCCN) and the American Association of Cancer Research (AACR) is provided below. Since this is a complicated disease affecting a wide variety of organs, it is recommended that individuals with MEN Type 1 be managed by a multidisciplinary team with expertise in medical genetics and the care of patients with this condition.

MEN1 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Pancreatic Neuroendocrine Tumors (PNET)	Lifetime risk ^{1, 3, 4, 5}	Up to 80%	<0.1%
Other <i>MEN1</i> -associated tumors and cancers	To age 80 ^{1, 3, 4}	Up to 100%	NA

MEN1 Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Pancreatic Neuroendocrine Tumors (PNET)	Clinical screening for symptoms ^{1, 4}	10 years	Individualized
	Biochemical screening ^{1, 4}	10 years	Annually
	Consider abdominal/pelvic CT or MRI with contrast ^{1, 4}	10 years	Every 1 to 3 years
	Consider serial endoscopic ultrasound ^{1, 4}	Individualized	Individualized
Other <i>MEN1</i> -associated tumors and cancers	Multiple screenings recommended, including clinical screening for symptoms, biochemical screening, and imaging with MRI, CT, ultrasound and other methods ^{1, 4}	Some screenings are recommended to begin as early as age 5	Variable by procedure

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the *MEN1* gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

Genetic testing should be considered for children of a parent known to have MEN type 1 by age 5, since some screenings begin at that age. If genetic testing is not done for children or other close relatives at risk for MEN type 1, they should be screened for complications of the condition beginning at age 5.⁴

Approximately 10% of individuals with MEN type 1 have not inherited the *MEN1* mutation from a parent. In these cases the mutation has developed spontaneously in that individual (a de novo mutation). Once this occurs, the children of that individual are each at 50% risk of inheriting the mutation.³

References

1. Bergsland E, et al. NCCN Clinical Practice Guidelines in Oncology®: Neuroendocrine and Adrenal Tumors. V 2.2022. Dec 21. Available at <https://www.nccn.org>.
2. Dreijerink KM, et al. Breast-cancer predisposition in multiple endocrine neoplasia type 1. N Engl J Med. 2014 371(6):583-4. PMID: 25099597.
3. Giusti F, et al. Multiple Endocrine Neoplasia Type 1. 2022 Mar 10. In: Adam MP, et al., editors. GeneReviews® [Internet]. PMID: 20301710.
4. Wasserman JD, et al. Multiple Endocrine Neoplasia and Hyperparathyroid-Jaw Tumor Syndromes: Clinical Features, Genetics, and Surveillance Recommendations in Childhood. Clin Cancer Res. 2017 23:e123-e132. PMID: 28674121.
5. SEER*Explorer: An interactive website for SEER cancer statistics [Internet]. Surveillance Research Program, National Cancer Institute. [Cited 2023 Mar 24]. Available from <https://seer.cancer.gov/explorer/>.

Last Updated on 31-Jan-2024